ESID/ERN-RITA Workshop
May 30-31 2022

FROM HIGH THROUGHPUT SEQUENCING TO DIAGNOSIS IN IMMUNE MEDIATED DISORDERS
Imagine Institute, 24 Boulevard du Montparnasse, Paris, France

**8:00-8:30 Participant welcoming**

Marielle van Gijn (Chair of the ERN RITA Molecular Testing Working Group) and Anne Puel (Chair of the ESID Genetics Working Party)

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**DAY 1**

**FOCUS ON 'GENOME' ANALYSIS INCLUDING INNOVATIVE MOLECULAR AND INFORMATICS TOOL RESEARCH AND DIAGNOSTIC METHODOLOGIES**

**8:30-10:15**

**NGS in IEI identification:** **PR ISABELLE MEYTS** (Laboratory of Pediatric Immunology, Department of Microbiology and Immunology, UZ Leuven, Belgium)

**From WES to WGE:** **DR AURÉLIE COBAT** (Laboratory of Human Genetics of Infectious Diseases, Imagine Institute, Paris, France)

**Long-read sequencing and human diseases:** **DR ALEXANDER HOISCHEN** (Lab of Genomic technologies & immunogenomics, Radboud University Medical Centre, Nijmegen, the Netherlands)

**10:15-10:45 BREAK**

**10:45-12:45**

**Mosaicism:** **DR ANNA MENSÀ** (Lab of Immunogenetics of the autoinflammatory response, Hospital Clínic de Barcelona, Barcelona, Spain)

**Clinical Genome resources, ClinGen, variant interpretation guidelines:** **PR JANNA SAARELA** (Institute for Molecular Medicine, Helsinki Finland)

**In silico programs as tools for identifying diseases causing variants:** **DR YUVAL ITAN** (Lab of Human disease genomics and computational biology, Mount Sinai, New York, US, online)

**Epigenetics and immune disease: DNA methylation:** **DR ESTEBAN BALLESTAR** (Lab of Epigenetics and Immune Disease, Josep Carreras Research Institute (IJC), Barcelona, Spain)

**13:00-14:00 LUNCH**

**14:00-15:30**

**Human Phenotype Ontology - HPO:** **DR MARIELLE VAN GIJN** (Lab of Genome diagnostic, Department of Genetics, University Medical Centre, Groningen, the Netherlands)

**Identification and study of Copy number variation (CNV):** **DR JÉRÉMIE ROSAIN** (Laboratory of Human Genetics of Infectious Diseases, Imagine Institute, Paris, France)

**Somatic revertants:** **DR ROGER COLOBRAN** (Lab of Translational Immunology Research Group, Immunology Division / Genetics Department, Universitat Autònoma de Barcelona, Barcelona, Spain)

**Gemma database: a new database for curation and search for variants, and high dimensional data in immune relevant genes:** **DR MICHELE PROIETTI** (Center for Chronic Immunodeficiency, University of Freiburg Medical Center, Freiburg, Germany)

**15:30-16:00 BREAK**

**16:00-18:00**

Case presentations: young fellows.
FUNCTIONAL VALIDATION STRATEGIES AND EVALUATION OF VUS – PATIENTS’ PERSPECTIVE

8:30-10:30
Monogenic Inflammatory Bowel Disease - genomics and variant validation in clinical practice: PR HOLM UHLIG
(Translational Gastroenterology Unit, University of Oxford, Oxford, UK)

Challenging cases: PR SOPHIE HAMBLETON (Translational and Clinical Research Institute, Newcastle University, Newcastle upon Tyne, UK), DR GIGLIOLA DI MATTEO (Bambino Gesù Hospital, Rome, Italy); DR KIMBERLY GILMOUR (Great Ormond Street Hospital, London, UK); DR VANESSA SANCHO-SHIMIZU (Imperial College, London, UK)

New Born Screening: DR MIRJAM VAN DER BURG (Laboratory for Immunology, Leiden University Medical Center, Dept. of Pediatrics, the Netherlands)

10:30-11:00 BREAK

11:00-13:00
Case presentations by young fellows.

13:00-14:00 LUNCH

14:00-15:00
Patients’ perspectives: JULIE POWER (Patient Contact and Policy Officer at Vasculitis Ireland Awareness)

Genetic diseases & Genomics, Genetic Counselling: DR MARCO CRIMI (Executive director Kaleidos, Bergamo, Italy)